Case #47

NAME Educational Activities Committee

Case provided by:

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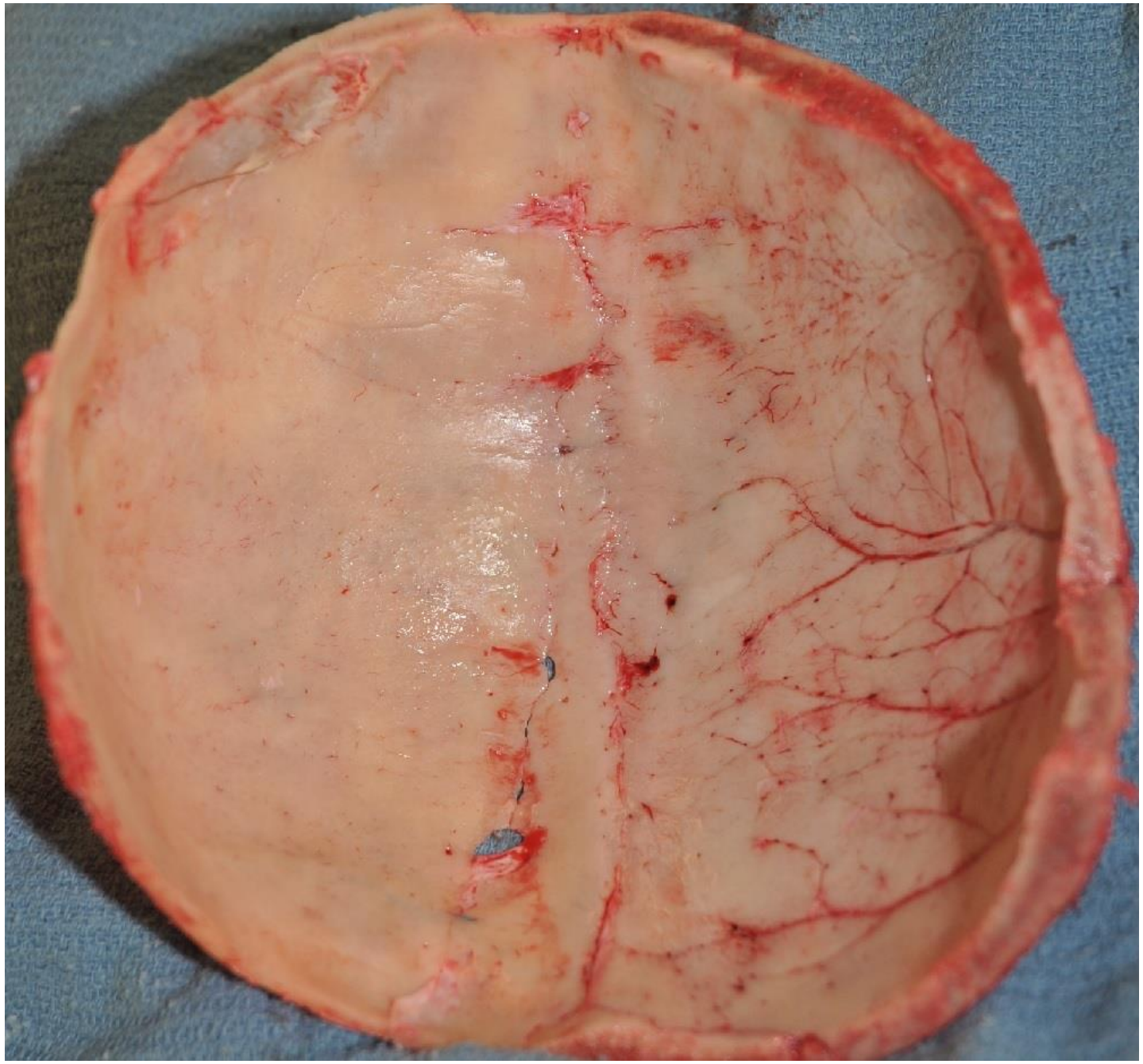

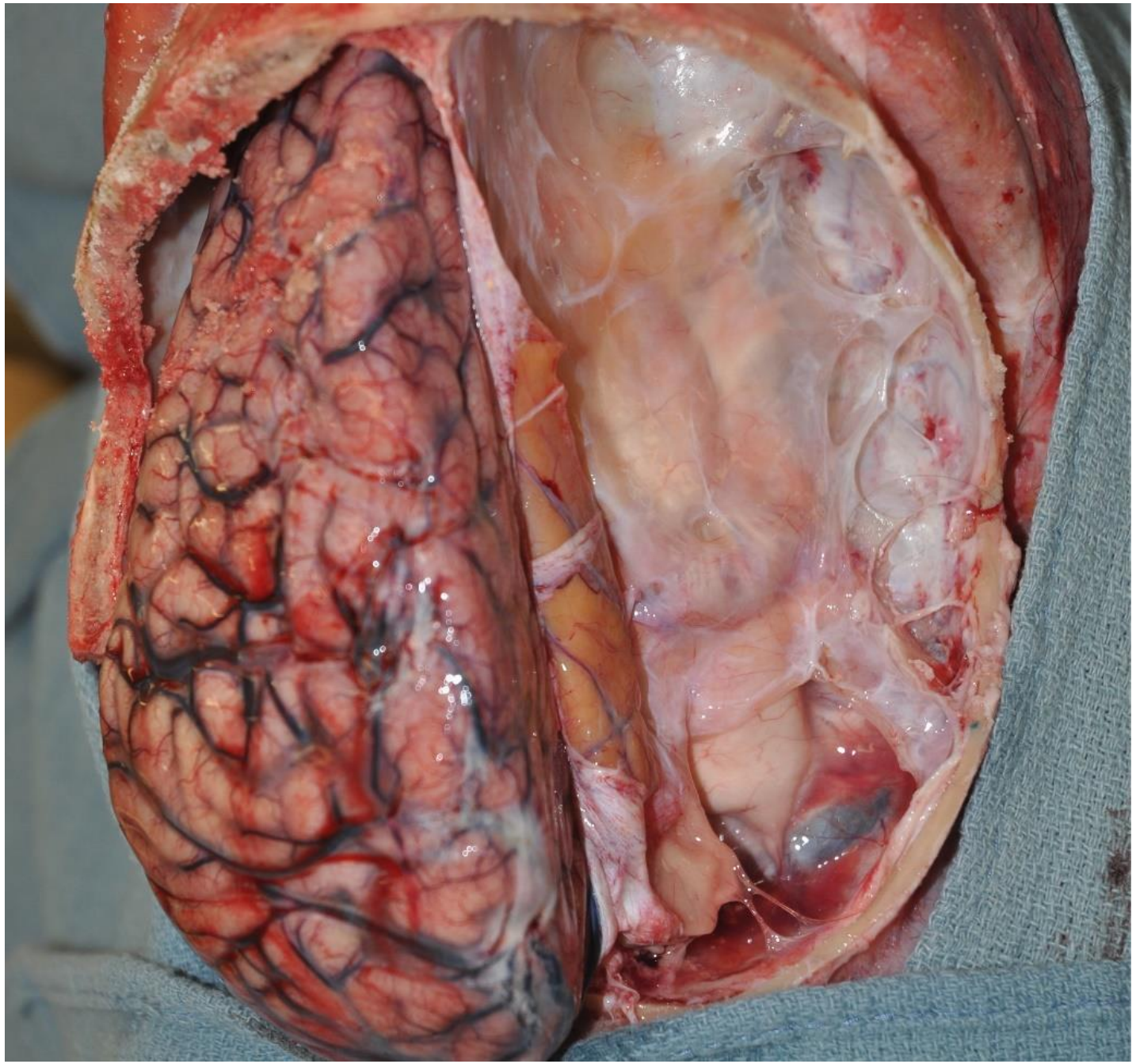

1. The images show the brain and calvarium of a 17-year-old girl who was found dead in her bed. She had a history of epilepsy, diagnosed when she was seven years old. She was reportedly appropriately taking anti-epileptic medications. No competing cause of death was identified at autopsy, and death was certified as Sudden Unexpected Death in Epilepsy (SUDEP). Which of the following best explains the finding seen in this picture?

- ◯ Therapeutic surgical intervention
- ◯ Congenital absence of right cerebral hemisphere
- ◯ Sequelae of remote ischemic event
- ◯ Childhood cerebral hemi-atrophy
- ◯ Post-traumatic hemi-cerebral degeneration

Answer...

A. Therapeutic surgical intervention (CORRECT ANSWER, 24.73 % of responses)

The picture above shows absence of the right cerebral hemisphere. The decedent on this case had a history of uncontrolled epilepsy due to Rasmussen's encephalitis. Rasmussen's encephalitis is a rare chronic inflammatory disorder that usually only affects one side of the brain. Patients often present with seizures, hemiparesis, and difficulty speaking.

In severe cases in which seizures are uncontrollable and determined to be coming exclusively from one side of the brain, a hemispherectomy (removal of one half of the brain) can be performed (note the remote surgical defects on the parietal bone picture). Interestingly, this radical surgery generally has a good outcome; 63% of patients in one study were seizure-free 5 years or more after surgery. The vast majority could also walk independently, and more than two-thirds could sufficiently carry on a conversation at follow-up.[1] However, seizures can persist even after surgery. In recent years, "functional hemispherectomy" (where the offending hemisphere is disconnected from the rest of the brain) has replaced complete anatomical hemispherectomy, as it results in similar outcomes with less morbidity.

The absence of endocalvarial vascular grooves is also important to note, indicating the chronicity of the condition, as the endovascular grooves form over time due to the blood pressure within the dural vasculature, in the setting of pressure inside the head that pushes the dura against the endocalvarium.

Sudden Unexpected Death in Epilepsy (SUDEP) is an investigative and diagnostic challenge for both death investigators and medical examiners. These deaths are diagnoses of exclusion, leaving non-specific findings, and can be misclassified as other types of natural deaths in the absence of a thorough medical history, autopsy, and death investigation. SUDEP can occur despite therapeutic anti-seizure medication levels. As such, it is thought that epilepsy-related deaths are significantly undercounted.

In 2018, members of NAME, clinicians, and researchers jointly produced recommendations for the investigation and certification of deaths related to epilepsy.[2] The goal was to raise awareness of SUDEP and produce consistency in how these deaths are certified. This greatly assists epidemiologists and researchers who study SUDEP to develop ways to monitor and treat epilepsy. The position paper also provides a tool for investigators to use in the field to help document important details in possible epilepsy-related deaths.

There are many ways to certify cases such as this, but certifiers are encouraged to use the word “epilepsy” and also emphasize known underlying etiologies. This death could be certified as “SUDEP due to Rasmussen’s encephalitis” or “epilepsy due to Rasmussen’s encephalitis.” Note, certifying the death as “complications of Rasmussen’s encephalitis” or just “seizure disorder” would make it more challenging for epidemiologists to identify this death as an epilepsy-related death. The 2022 NAME Interim Meeting will have two lectures on neuropathology of pediatric epilepsy.

B. Congenital absence of right cerebral hemisphere (35.33% of responses)

There are rare documented cases of congenital absence of one hemisphere of the brain, however, the defects in the parietal bone are more consistent with prior surgery.

C. Sequelae of remote ischemic event (6.91% of responses)

While it is possible for remote ischemic events to only affect one hemisphere, it is more common for them to affect both hemispheres in vascular distributions. Depending on when the ischemic insult occurred, it may manifest in several different ways including “basket brain”, porencephaly, hydranencephaly, multicystic encephalopathy, and global hemispheric necrosis. If the ischemic event occurs later in life (after the perinatal period) presentation may vary depending on the vascular distributions affected.

D. Childhood cerebral hemi-atrophy (23.35% of responses)

Cerebral hemi-atrophy (CHA) is uncommon and can have many etiologies (intrauterine vascular injury, birth trauma, perinatal intracranial hemorrhage, Rasmussen encephalitis, prolonged febrile seizures, infections, vascular abnormalities, neoplasia, radiation, and others). The congenital types usually occur prenatally and acquired types generally occur in the first two years of life. CHA typically leaves some residual remnant of brain tissue in the involved hemisphere. Additionally, when CHA occurs, it causes changes in the calvarium including ipsilateral calvarial thickening, diploe widening, hyper-pneumatization of paranasal sinuses/mastoids, and elevation of petrous bone and middle cranial fossa [3].

E. Post-traumatic hemi-cerebral degeneration (9.68% of responses)

There is no specific entity called “post-traumatic hemi-cerebral degeneration,” and it would be unlikely for any sort of remote trauma to present like this without surgical intervention.

REFERENCES

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